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Chapter

Ultrasonic Detection of Down Syndrome Using Multiscale Quantiser with Convolutional Neural Network

Michael Dinesh Simon and A.R. Kavitha

Abstract

Down Syndrome is a genetic condition that occurs when there is an extra copy of a chromosome 21 in the newly formed fetus. EIF is observed as one of the possible symptoms of DS. But in comparison to the other symptoms like nasal bone hypoplasia, increased thickness in the nuchal fold, EIF is very much less prone to DS. Hence, recommending the pregnant women with EIF to undergo the diagnostic process like amniocentesis, CVS and PUBS is not always a right choice as these diagnostic processes suffer serious drawbacks like miscarriage, uterine infections. This chapter "Ultrasonic Detection of Down Syndrome Using Multiscale Quantiser With Convolutional Neural Network" presents a new ultrasonic method to detect EIF that can cause DS. Ultrasonic Detection of Down Syndrome Using Multiscale Quantiser with Convolutional Neural Network entails two stages namely i) training phase and ii) testing phase. Training phase aims at learning the features of EIF that can cause DS whereas testing phase classifies the EIF into DS positive or DS negative based on the knowledge cluster formed during the training phase. A new algorithm Multiscale Quantiser with the convolutional neural network is used in the training phase. Enhanced Learning Vector Classifier is used in the testing phase to differentiate the normal EIF from EIF causing DS. The performance of the proposed system is analysed in terms of sensitivity, accuracy and specificity.

Keywords: echogenic intracardiac focus, down syndrome, cross-correlation, enhanced learning vector quantiser

1. Introduction

Down syndrome (DS) is the chromosomal abnormality caused in humans when extra genes from chromosome 21 are transferred to a newly produced embryo. It affects the fetal development leading to physical and mental abnormalities. The babies with DS have a distinct appearance than normal babies. Some of the DS victims are shown in **Figure 1**. The babies suffering from DS are likely to have retardation in growth and the mental problems. DS, in general, cannot be cured; the simplest way to avoid the babies with DS is to detect the fetus with DS and prevent it from being born.

DS is not a rare phenomenon and the occurrence of this phenomenon is eventually improving throughout the world. Patterson [1] in his work "Molecular genetic

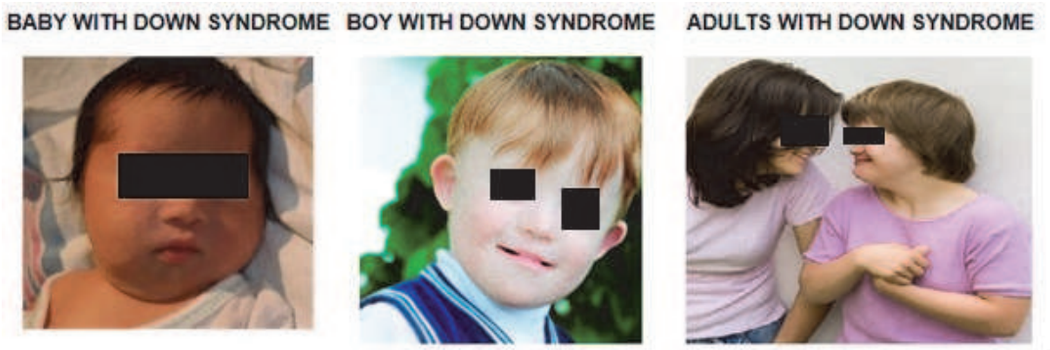

Figure 1.
Victims of DS (courtesy: www.womens-health-advice.com).

analysis of Down Syndrome" states that more than 1 in 1000 neonates has DS. A stats report [2] shows that i) the risk of DS in the global birth rate is 0.00125% ii) women in the age of 30 and below have 0.001% risk of DS iii) women in the age of 45 have 0.022% risk of DS. The abortion rates in DS-affected pregnancies [3] have amplified to 67–92% in the United States and Europe.

In medical world, ultrasonographic markers like nuchal fold, nasal bone hypoplasia and femur length are often seen as a symptom of DS. Nuchal fold is a skin fold noted at the backside of neck in the fetus during the second trimester. Increased thickness in the nuchal fold is observed as the most sensitive symptom of DS. A comparison of the nuchal fold between the normal fetus and DS affected fetus is shown in **Figure 2**.

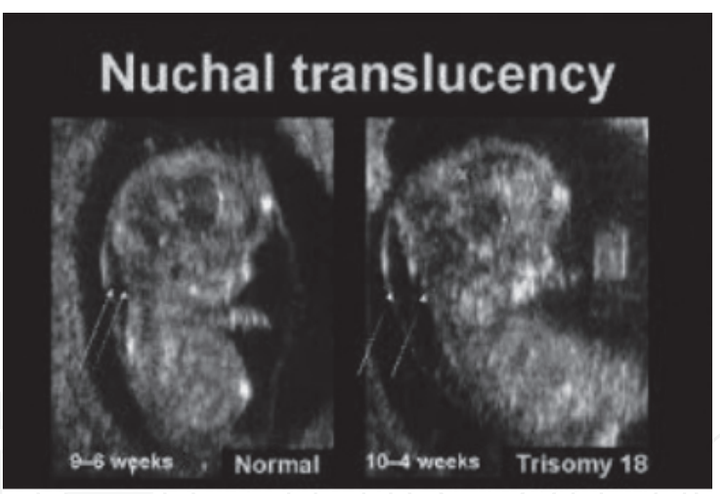

Figure 2.
Comparison of nuchal fold between normal fetus and DS related fetus (courtesy: http://babysmith2014.blogspot.com).

Nasal bone hypoplasia is a condition where the nasal bone of the fetus appears to be very small. It has to be noted that 70% of DS [4] fetuses have no nasal bone or smaller nasal bone. A comparison of nasal bone analysis between the normal fetus and DS related fetus is shown in **Figure 3**.

Femur length is the measure of the longest bone in the fetus. Shortening of femur length is considered as the symptom of DS. Recently, Echogenic Intracardiac Foci (EIF) had been considered as a new symptom of DS. Many clinical researches affirm that the presence of EIF is considered as a potential indication of DS. An EIF is named as an intense white shape that is found in the heart of the fetus. It can be examined through the ultrasound. An article "Ultrasound Findings" expresses that EIF is seen in around 1 out of each 20 or 30 pregnancies (~3–5%). EIF is associated with 12% of foetuses with DS [5] and biventricular EIF suffers higher risk for

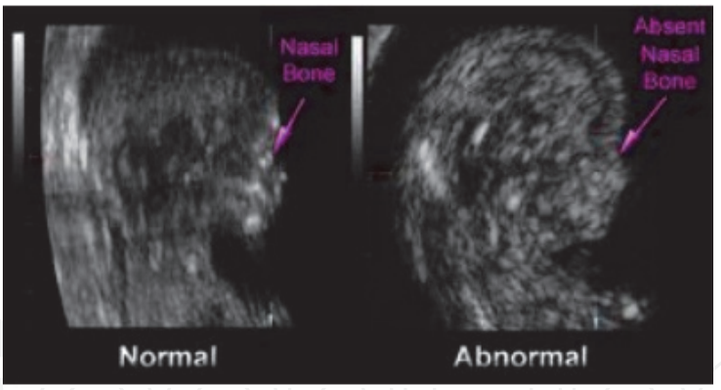

Figure 3.
Comparison of nasal bone between normal fetus and DS related fetus (courtesy: http://babysmith2014.blogspot.com).

aneuploidy. If an EIF is detected in an ultrasound image, the pregnant women with EIF fetus feel dreadful mental pressure fearing that they would deliver DS affected baby [2]. This psychological weight caused in the pregnant ladies can undesirably affect the fetus. To cut the down the psychological weight of the pregnant ladies and to guarantee that the fetus is unaffected with DS, women with EIF symptoms are prescribed to go through chorionic villus sampling and amniocentesis.

Amniocentesis, Percutaneous umbilical blood inspecting and Chorionic villus sampling methods are the most non-ultrasonic method for detecting DS. Amniocentesis is a method of collecting a little amount of the amniotic fluid that surrounds the fetus and analysing it for trisomy 21. It is carried in the light of ultrasonic guidance. It is advised after the 15th week of pregnancy. This method has the high risk of leaking amniotic fluid, miscarriage, needle injury to fetus and infection transmission. Percutaneous umbilical blood sampling (PUBS) is a method of collecting the blood from the umbilical cord and tests it for chromosomal defects. It is performed in the 18th week of the pregnancy. PUBS have a higher risk of miscarriage. Chorionic villus sampling (CVS) is a method of analysing the chromosomes in the cells taken from the placenta. CVS is performed between 9th and 14th weeks of the pregnancy. This method suffers from the drawbacks like miscarriage and uterine infection problems.

As the relativity of EIF with DS is very low, it would not be advisable to request women with EIF fetus to undergo Amniocentesis, CVS and PUBS. There is a great need to coin a new mechanism that can differentiate DS related EIF fetus from the normal EIF fetus through ultrasound testing. This research aims at bringing a new invention that can detect DS based on EIF through an ultrasound scan.

The major contributions of this chapter are:

- A new medical parameter EIF is used for the detection of DS
- A new image segmentation algorithm Multi-scale Quantized Convolution Neural Network (MSQCNN) is developed and used for accurately detecting and extracting the EIF in the ultrasound fetal images
- Cross-Correlation Technique (CCT) is employed to confirm DS by analysing the nasal bone hypoplasia in the training phase
- A new supervised classification scheme Enhanced Learning Vector Quantiser (ELVQ) is utilised to differentiate the normal EIF from the EIF related to DS.

Introduction of the problem statement, the necessity for this research and the challenges present in this research had already been well discussed in section 1. The

rest of this chapter is structured as follows. A complete view of related studies is provided in section 2. The proposed methodology is elucidated in section 3. The test results and comparisons are elaborated in section 4. Future research directions and conclusion are briefed in section 5 and section 6.

2. Related works

This section provides a detailed view of the emerging researches performed on the identification of DS utilising ultrasound. Rebecca et.al [6] performed analyses to decide the exactness of second-trimester ultrasound in identifying DS in fetuses. The malformation in the structure and ultrasonographic markers are the baseline factor behind this research examination. The experimental outcomes demonstrated that the ultrasonographic markers without related morphological abnormalities could not differentiate between the normal fetus and the fetus with DS. This research made a noteworthy observation that placing the markers as a deciding factor to suggest amniocentesis can lead to a huge number of fetal losses.

Lauren Lynch et.al [7] brought forth the new ultrasonic parameter nuchal fold that was highly informative for detection of DS than the other parameters like biparietal diameter and femur length. Patrick Rozenberg et.al [8] recommended the inclusion of the first-trimester ultrasound scan for the diagnosis of DS in addition to the second-trimester ultrasound. Detection of the ultrasonographic marker like nuchal fold, biparietal diameter and femur length is a challenging task due to the presence of speckle noise in an ultrasound image. Detection of these markers requires highly skilled sonologists, obstetricians, and fetal medicine professionals [2]. This led to the paradigm shift for the automated diagnosis of DS from the ultrasound images.

Cuckle et al. proposed a genetic scan mechanism called nuchal translucency scan [9] to assess the risk of the fetus suffering from DS based on the nuchal fold. NT scans works based on the principle that “fetus with DS will accumulate more fluid at the end of the neck”. NT scan is always performed between 11 to 13 weeks of the pregnancy. As the risk of Aneuploidy and DS increases exponentially with the thickness of NT thickness [10], there has been wide growth in the mechanism of semi-automated methods for detecting DS based on the nuchal fold.

Yinhui Deng et al. [11] have presented a systematic structural model for the automated detection of nuchal translucency region. Anzalone et al. [12] automated the measurement of nuchal translucency from the ultrasound image. Nirmala. S and Palanisamy. V [13] proposed the identification of nuchal translucency utilising the imaging methods like mean shift analysis and canny operators. The thickness of nuchal translucency was computed utilising blob analysis. Sonia. R and Shanthi V [14] performed the morphological operations and Otsu thresholding for the partitioning and calculating the region of nuchal translucency.

Lai K Wee et.al [15] utilised the neural network to figure out the area of nuchal translucency. The boundary region of nuchal translucency layer is detected utilising an instinctive computerised algorithm. After the identification of the boundary region, the optimum thickness of the region is computed based on intensity continuity and edge strength. Yan Du et.al [16] performed fetal karyotype assessment among Chinese ladies and found that nonappearance of nasal bone in the second trimester has the high risk of DS in the fetus. Iliescu Dominic-Gabriel and Drăgușin Roxana-Cristina [17] reviewed various schemes in early hereditary screening and ultrasound assessment and inferred a decision that first-trimester ultrasound assessment can possibly analyse the most of fetal irregularities.

Shafia Shakoor, Humera Ismail and Shama Munim [18] explored the experimental results in fetuses with EIF. The trial was experimented in Pakistan and the outcomes showed that 95.77% of the fetuses were normal, just 4.2% recorded abnormalities in heart and 0% had DS. Aaron B. Caughey, Deirdre J. Lyell et.al [19] assessed the impact of isolated EIF for screening DS. The investigation affirmed the use of EIF as a screening factor may prompt an immense number of amniocenteses and miscarriages to decide a rare DS fetus.

The following observations are evident from the recent researches carried out in this field I) The most often utilised parameters for the identification of DS are nuchal fold, femur length and nasal bone hypoplasia ii) Association of DS and EIF is relatively low iii) There exist a need and necessity for developing a computerised methodology for spotting DS, as the manual diagnosis requires some artifacts.

3. Proposed methodology

Histopathology is the assessment of tissues in the impacted organs of the body. The extensive growth of the computerised image processing has given rise to a new diagnostic methodology termed as computer-assisted diagnosis (CAD). CAD assists the radiologists for disease detection, diagnosis and prognosis prediction. Diagnosis of DS based on EIF is one of the difficult issues that could be tackled by CAD methodology. The efficiency of imaging techniques and CAD can be unitedly employed to substitute the traditional DS diagnostic methods like amniocentesis and chronic sampling.

The proposed system "Ultrasonic Detection of Down Syndrome using Multiscale Quantiser with Convolution Neural Network" uses a medical parameter EIF for the detection of DS. It is designed with the aim of discriminating the normal EIF from DS related EIF. It consists of two phases i) training phase and ii) testing phase. Training phase involves learning and forming the knowledge cluster of EIF related to DS. Testing phase involves classifying the diagnostic ultrasound fetal image as DS positive or negative, based on the knowledge cluster. An architecture diagram of the proposed system is given below in **Figure 4**.

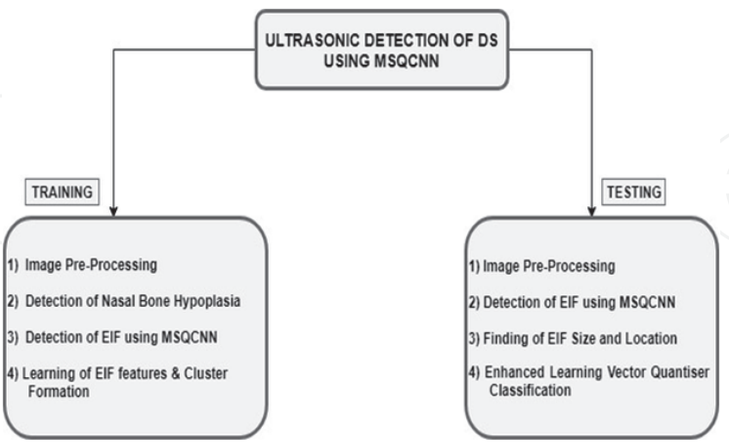

Figure 4.
The architecture of the proposed system.

3.1 Training phase

Not all the fetus that contain EIF result in DS, only few patterns of EIF can cause DS. The aim of this phase is to find the characteristics of DS-related EIF. The training phase includes five steps i) Image Pre-Processing using Neuro-Fuzzy Filter – to eliminate the speckle noise in the ultrasound fetal image that can hinder the

detection of EIF ii) Detection of Nasal Bone using CCT - to confirm the presence of DS in the ultrasound fetal image iii) Detection of EIF using MSQCNN – to detect/segment EIF present in the ultrasound fetal image iv) Learning the features of EIF and cluster formation – to analyse the features of EIF like size, location and form a cluster. The systematic representation of the training phase is shown in **Figure 5**.

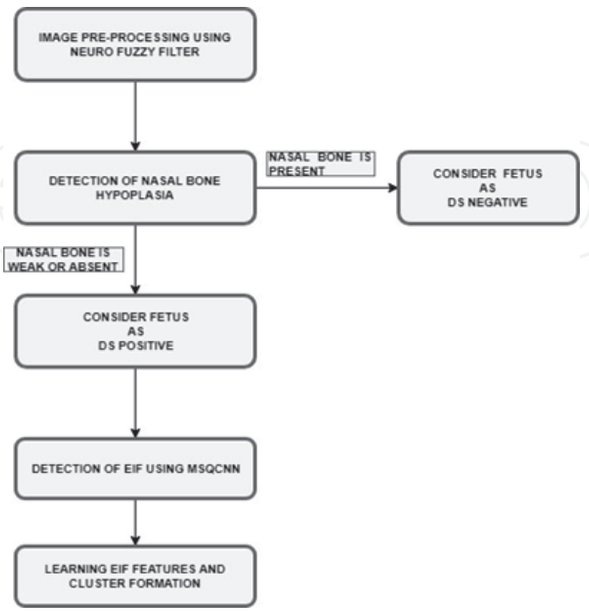

Figure 5.
A schematic representation of the training phase.

3.1.1 Image pre-processing

The ultrasound devices produce high quality, real-time images but very often it is degraded with speckle noise. Speckle noise is a big threat for the quality of the results of image processing. It can produce artificial edges, echoes the patterns in the images that disturb the diagnosis process.

The image pre-processing is the most significant step of the training phase. In pre-processing; a neuro-fuzzy filter that operates based on neuro-fuzzy and evolutionary learning is used to eliminate the speckle noise. The neural network possesses the ability to train. The knowledge for the training is fed into the system in a fuzzy format. Speckle noise is eliminated based on the fuzzy knowledge. A memetic algorithm is utilised to improve neuro-fuzzy filter. The algorithmic description of neuro-fuzzy filter is given below:

Algorithm: Neuro-fuzzy filter
Input: Ultrasound image with speckle noise
Output: Processed image with less noise
Algorithm

- For each ultrasound image X1, there presents 25 membership function (μ)
- Membership function can generate 25 transitional outcomes Y1, Y2....Y25
- Find the weighting factor for each individual membership
- Average the weight of the individual rule's output and form Yo output.

3.1.2 Detection of nasal bone using CCT

Detection of nasal bone is the second step of the training phase. The presence of DS is confirmed by the absence of nasal bone using CCT, as seventy percent of DS

foetuses have no nasal bone [4]. CCT is a simple matching algorithm that works based on the correlation. The algorithmic description of CCT is displayed below [20].

Procedure: Cross correlation technique
Input: Pre-processed image
Output: 3D surface plotted graph
Algorithm

1. Start
2. Consider target image S and template image T.
3. Consider the ultrasound image S for which nasal bone has to be detected

$$S'i,j = \frac{1}{mxn} \sum_{i=0,j=0}^{n-1,m-1} S(i+x, j+y) \quad (1)$$

where Si → subset of the target image,
i and j are coordinates of the template image
x and y are coordinates of the target image

4. Consider the ultra sound image T with nasal bone detected

$$T' = \frac{i}{mxn} \sum_{i=0,j=0}^{n-1,m-1} Ti,j \quad (2)$$

where T′ → subset of template image

5. Derive subset image(I) in T image that include nasal bone area
6. Look for I in S image
7. If I is present in S, obtain the location of I
8. Find cross correlation matrix for the template image and target image

$$\partial = \frac{J(Si+x,\ j+y-S'i,j)(Tx,y-T')}{\sqrt{J(Si+x-S'i,j)2 * J(Tx,y-T')2}} \quad (3)$$

where $J = \sum_{x=0,y=0}^{n-1,m-1}$ -> summation of co-ordinates in the template image

9. The correlation matrix is converted into surface plotting graph
10. Based on the graph, the maximum value of image correlation will be used for the detection of nasal bone
11. If the peak of the graph is less than 0.35, it can be concluded that nasal bone is absent
12. Stop

3.1.3 Detection of EIF using MSQCNN

Detection of EIF is the third step in the training phase following the image pre-processing and detection of the nasal bone. Training phase continues to the detection of EIF, only when the detection of nasal bone reports that fetal image has no nasal bone or weak nasal bone in it.

MSQNN is used to detect the EIF in the ultrasound images. MSQNN consists of additional components such as acceptor, quantiser with the conventional five layers that include convolution layer 1 and 2, pooling layer 1 and 2, fully connected layer 1 and 2. The output object coming out of FC Layer 2 is led into acceptor. The functionality of acceptor is to compare the object detected in the various scale of the input image and compute the difference between the objects. If the difference between the objects detected at the various scale lies within the threshold, it is treated as the final object. Acceptor computes the difference between the objects using the following formula.

$$\partial = \frac{1}{n} \in \quad (4)$$

where ∂ = Difference between the object detected at scale 1 and scale 2,
Y (i) = Object detected at scale 1,
Y (i + 1) = Object detected at scale 2,
SF = Scale factor at which object is detected.

Acceptor stops forwarding the control to Quantiser if the acceptance criteria are met. Below is the condition for the acceptance criteria to be met.

$$\partial t < = \partial \tag{5}$$

where ∂t = threshold difference between objects detected at various scale (specified by the user).
∂ = actual difference between objects detected at various scale.

If the acceptance criterion is not met, acceptor transfers the control to quantiser. Quantiser performs quantisation by dividing the input image by scaling factor and rounding this value to the nearest integer. The mathematical formulation of quantiser is presented below.

$$X'(u,v) = Round\frac{X(u,v)}{Q(u,v)} \tag{6}$$

The next iteration for object detection starts with the reduced size input image. This iterative process of detecting an object continues until the number of iterations reaches the maximum or the acceptance criterion is met in Acceptor. The final object detected is treated as the EIF present in the ultrasound image of the fetus.

The pictorial representation of Multiscale Quantised Convolution Neural Network is given below in **Figure 6**.

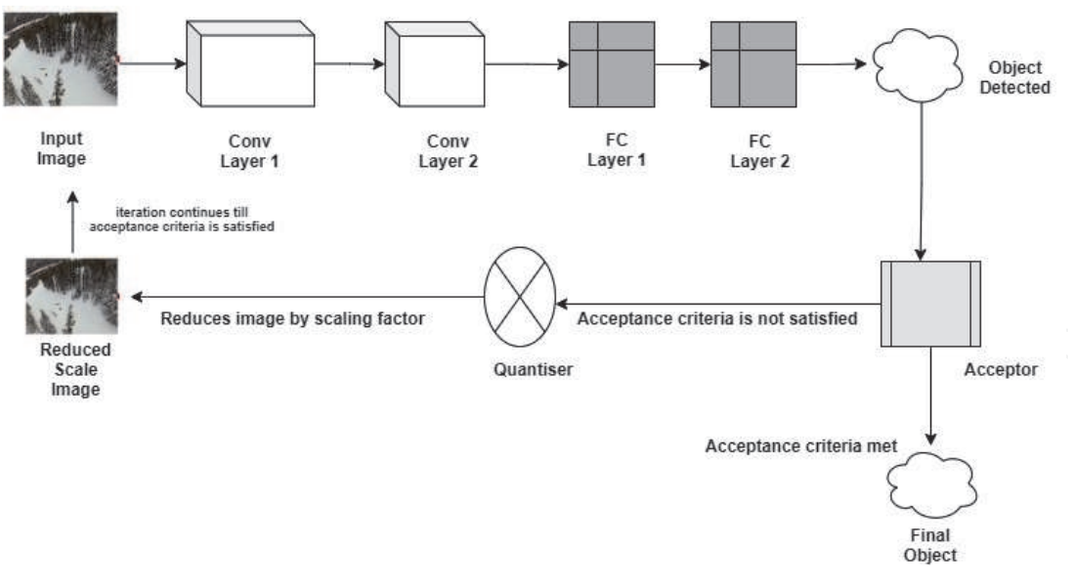

Figure 6.
A pictorial representation of multiscale quantised convolution neural network.

3.1.4 Learning the features of EIF

Learning the features of EIF is the last step of the training phase, it happens just when the nasal bone is absent. The size and spatial coordinates of EIF are stored and clustered based on the closeness of the appearance (2). The similarity parameter utilised for clustering are i) size of EIF and ii) position of EIF specifically left ventricle and right ventricle. These clusters become the knowledge base for ELVQ function of the testing stage. Basically, two clusters are formed in this

training phase of ELVQ, one cluster is for DS positive EIF fetus and other is for DS negative EIF fetus.

3.2 Testing phase

Testing phase involves discriminating DS related EIF from the normal EIF. The knowledge attained in the training phase plays the critical role in the determination of DS-related EIF. ELVQ maps the testing image to the cluster map developed in the training phase, by spotting the malignant EIF.

The testing phase contains five steps namely i) Image Pre-Processing using Neuro-Fuzzy Filter – to eliminate the speckle noise in the ultrasound fetal image that can hinder the detection of EIF ii) Detection of EIF using MSQCNN – to detect EIF present in the ultrasound fetal image iii) Find the features of EIF – to find the features of EIF like size and location iv) Enhanced Learning Vector Quantisation Classification – to classify fetus into DS positive or negative based on the knowledge cluster. The flowchart representation of the testing phase is shown in **Figure 7**.

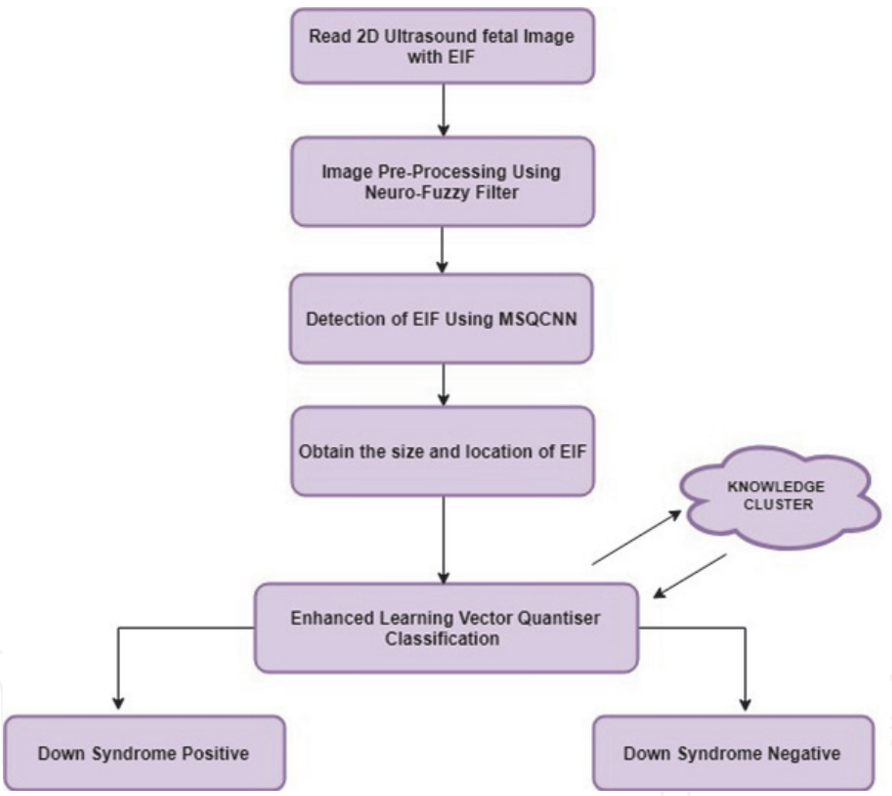

Figure 7.
A flowchart representation of the testing phase.

3.2.1 Enhanced learning vector Quantiser

The first two steps of testing phase are i) Image Pre-Processing and ii) Detection of EIF using MSQCNN are discussed well in the section 3.1.1 and 3.1.3. The third step concentrates on obtaining the features of EIF. Then as the fourth step, ELVQ classification involves.

ELVQ supervised learning classifies the test image into DS positive or negative groups. For a single testing, two steps of ELVQ should be finished. In the initial step, the size of EIF is treated as the weight vector and in the second the location of EIF is treated as the weight vector (2). Enhanced Learning Vector Quantiser differs

from the conventional Learning Vector Quantiser in one aspect; LVQ uses Euclidean distance whereas ELVQ uses Manhattan distance function.

Procedure: Enhanced Learning Vector Quantiser
Input: network weights, the learning rate, neighbourhood radius
Output: Classification of zp input vector [21]

1. Set the weights of network, the rate of learning and the neighbourhood radius;
2. while stopping condition(s) not true do
3. for each pattern p do
4. Compute Manhattan distance, dk,p, among input vectors zp and each
   Weight vector uk = (uk1, uk2, · · ·, uKI) as
   dk,p (zp,uk) = |zp – uk| (7)
5. Calculte the output unit ok for which the distance dk,p is the lowest;
6. Update all the neighbourhood weights κk,p using Eq. [7]
   end
7. Update the learning rate;
8. Reduce the neighbourhood radius at specified learning iterations;
9. End

At the end of this testing phase, ELVQ classifies an input EIF fetus image as DS positive or negative.

4. Results and discussions

This section describes the experimental results of the proposed techniques. The proposed system is implemented with software Scilab. It [2] was employed for image and mathematical processing needs. Scilab is open source and freeware that can perform wide numerical computation for engineering and scientific applications.

4.1 Dataset description

As there is no standard dataset for EIF fetus images, the experiment was carried out on the user created the dataset. Nearly 35 2D-ultrasound fetal images were collected from numerous online and offline sources. These fetal images belong to the period of 24–26 weeks. Out of these 35 fetal images, the diagnosis result of DS is known for 25 fetal images (that includes 20 ultrasound fetal images with DS positive and 5 ultrasound fetal images with DS negative).

These 25 ultrasound fetal images are used in the training phase to obtain the characteristics of EIF associated with DS. There were also 10 ultrasound fetal images with EIF with no clue for DS were used in the testing phase. Some of the fetal images with EIF that were used for the experiment were shown in **Figure 8**.

4.2 Experimental results

Training was performed with 25 DS diagnosed images. It was done in five iterations and the learning rate was consistently improving. The training phase results were presented in **Table 1**. The average rate of the learning at the end of training phase was 80%, while the rejection rate was only 20%.

Experimental results of the training phase are given as follows:

Detection of EIF using MSQCNN is shown in **Figure 9**.

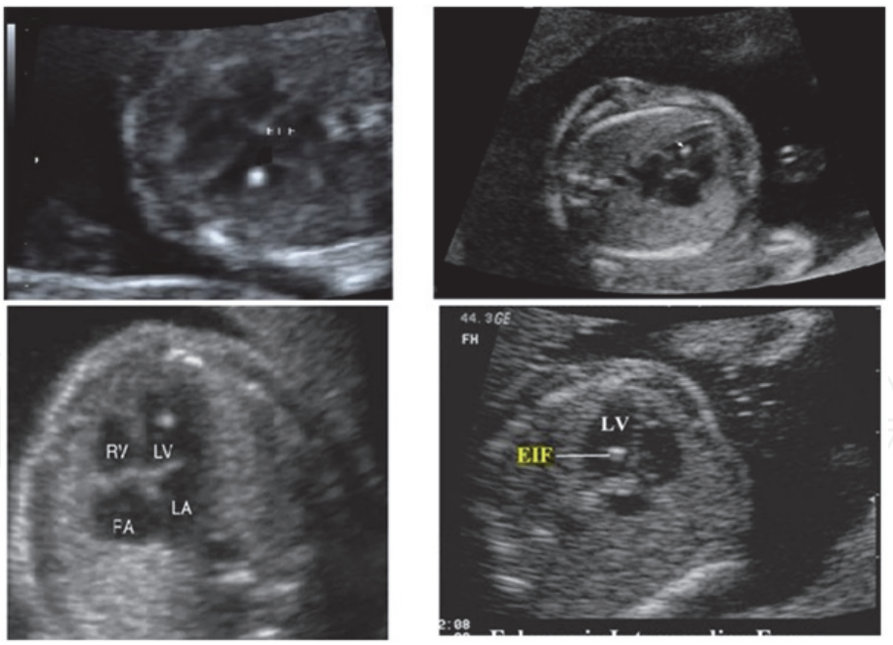

Figure 8.
The dataset of the fetal images with EIF.

Iteration No#	No. Of Input Images	Learning Rate	Rejection Rate
1	10	10	0
2	20	14	6
3	40	34	6
4	80	68	12
5	100	86	14

Table 1.
Training phase results.

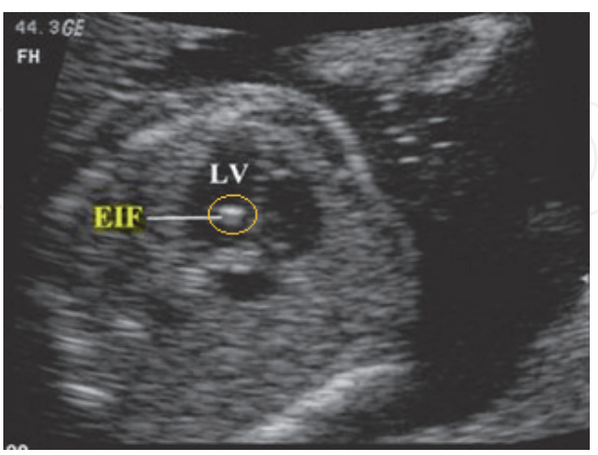

Figure 9.
Detection of EIF.

Detection of nasal bone using CCT in **Figure 10**.

The fetal image (A. Normal) that was rejected and the fetal image (B. Down syndrome) that was learnt in the training phase is shown in **Figure 11**.

The knowledge attained from the training phase about the location and size of EIF is shown in **Table 2**.

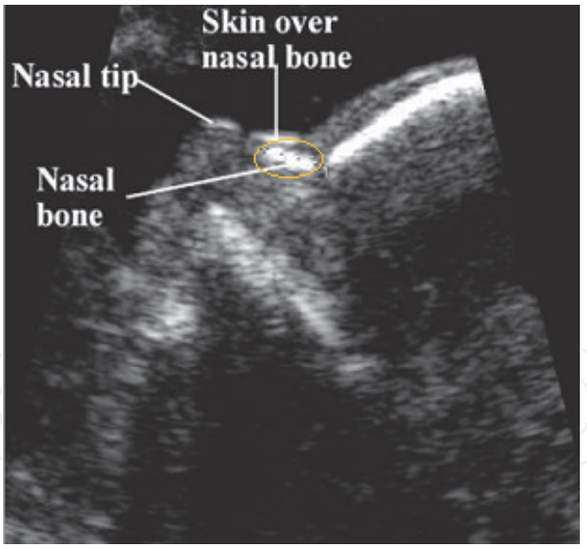

Figure 10.
Detection of nasal bone.

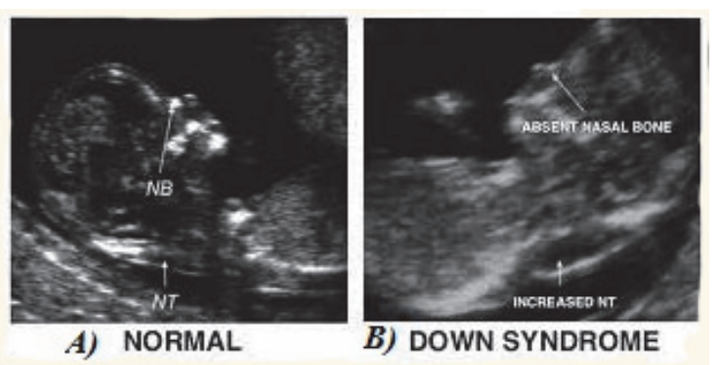

Figure 11.
Results of the training phase.

S.No	Size of EIF	Location of EIF	Other Ultrasound findings	DS Positive
1	> = 5.4 mm	Left Ventricle	Nasal Bone Absence	Yes
2	> = 5 mm	Biventricular	Nasal Bone Absence	Yes

Table 2.
Clustered knowledge from the training phase.

The testing phase was carried out to identify DS in 5 fetal images with EIF is shown in **Table 3**.

Experiment No #	Tested images	Down Syndrome Positive	Down Syndrome Negative
1	50	33	17

Table 3.
Results of the testing phase.

The fetal image with EIF that was analysed for DS in the testing phase and proved to be Down syndrome negative is displayed in **Figure 12**.

The fetal image with EIF that was analysed for DS in the testing phase and confirmed as Down syndrome positive is displayed in **Figure 13**.

Figure 12.
26 weeks fetus with left ventricular EIF with DS negative in the testing phase.

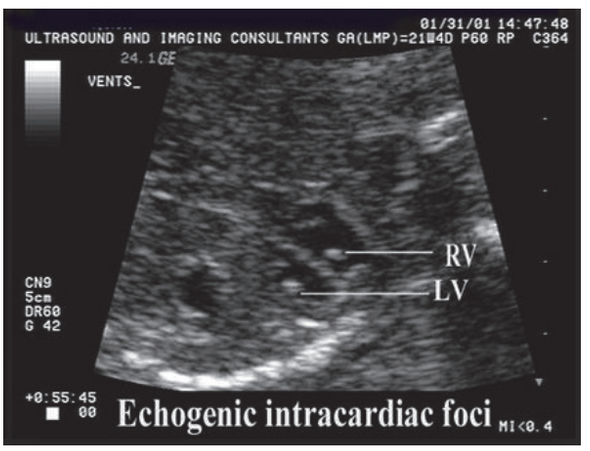

Figure 13.
A fetus with EIF on both the ventricles with DS positive in testing phase. X.

4.3 Performance analysis using evaluation metrics

The performance of DS detection system is evaluated using parameters such as sensitivity, specificity, and accuracy. Sensitivity is the number of true positives that are rightly acknowledged by a diagnostic test [2]. It states how well the diagnostic test is identifying a disease.

$$\text{Sensitivity} = \text{T.P}/(\text{T.P} + \text{F.N}) \tag{7}$$

Specificity is the quantity of the true negatives rightly acknowledged by a diagnostic test [2]. It determines how good the test is identifying normal (negative) condition.

$$\text{Specificity} = \text{T.N}/(\text{T.N} + \text{F.P}) \tag{8}$$

Accuracy is the number of true results, either true positive or true negative, in a population. It measures the degree of veracity of a diagnostic test on a condition.

$$\text{Accuracy} = (\text{T.N} + \text{T.P})/(\text{T.N} + \text{T.P} + \text{F.N} + \text{F.P}) \tag{9}$$

(Note: T.P stands for True Positive, T.N stands for True Negative, F.P stands for False Positive, F.N stands for False Negative)

The performance metrics of the proposed system is compared with the existing system "Automatic Computerized Diagnostic Tool for Down Syndrome Detection in Fetus" [2] is shown in **Figure 14**.

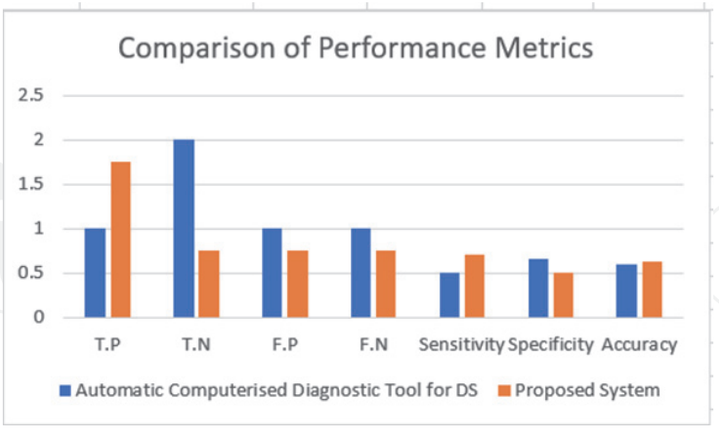

Figure 14.
Comparison with the state of art.

The proposed system showed the better performance than the existing system terms of sensitivity and accuracy, but it showed low specificity than the existing system.

5. Future research directions

The newly designed system was able to clearly differentiate DS related fetus from the normal fetus based on EIF. It was producing very accurate results when operated on the fetal ultrasound images with a single EIF and multiple EIF. In future works, the soft markers like nuchal fold and femur length can be considered as an alternate parameter instead of nasal bone hypoplasia in the training phase.

6. Conclusion

This chapter presented a new idea "Ultrasonic Detection of Down Syndrome using Multiscale Quantiser with Convolutional Neural Network" to detect DS based on EIF in an ultrasonic automated method. The proposed system was intelligent enough to clearly distinguish DS causing EIF from the normal EIF. It attained better results in terms of accuracy, sensitivity, and specificity. In future works, this system can be added as a new feature in the ultrasound fetal scan. It can also serve as an alternate for the conventional DS diagnostics like amniocentesis, PUBS, and CVS.

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